Short Atlas in Pediatrics

Spot Diagnosis of the most common Pediatric Diseases

Mohammed El-Naggar

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Mohammed El-Naggar

Professor of Pediatrics and
Pediatric Intensive Care
Faculty of Medicine, Cairo University
Cairo, Egypt

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Mohammed El-Naggar

25 Dokki Street, Cairo, Egypt
E-mail: mrnaggar45@hotmail.com

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Preface

Visual recognition of clinical signs is extremely important in clinical diagnosis. Visual memory is strong and long lasting, and one image is probably more effective than several pages.

In this short atlas, the most important clinical signs are collected in 8 chapters and in around 200 new images. With each image, description of the relevant findings and the important criteria for diagnosis are included.

Some important chapters as chest and cardiac diseases are intentionally omitted because diagnosis of these diseases depends mainly on hearing and not seeing.

In this short book every sincere effort has been made to make it as simple as possible and I hope it can be useful for medical students and young pediatricians.

Mohammed El-Naggar

2005

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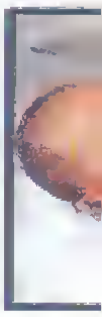

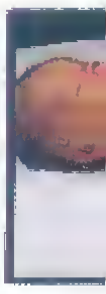

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NEWBORN

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5. Common umbilical lesions.
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1. Skin Color Variations

Normal pinkish appearance

The color of the normal newborn is reddish at birth and it changes to pink within a day or two.

However, the pink color at rest may be transformed to a deep red on crying.

Reddish color of premature

The color of the premature baby is deep pink to red. In severe prematurity (as opposite figure), the color is deep red. Note also the small size (compared to the examiner's hand) and the relatively large head.

Cutis marmorata

Cutis marmorata is a violaceuos or blue mottling of the skin that occurs when the infant is exposed to low temperature (as during bathing or changing). This lacy reticulated pattern appears over most of the body surface. It is an accentuated vasomotor response that disappears with age.

2. Neonatal Jaundice

Physiological jaundice

Physiological jaundice is a very common condition that occurs in up to 40% of normal newborns. It usually appears on the third day and remains for less than a week. In most cases, the bilirubin level is quite below the critical level.

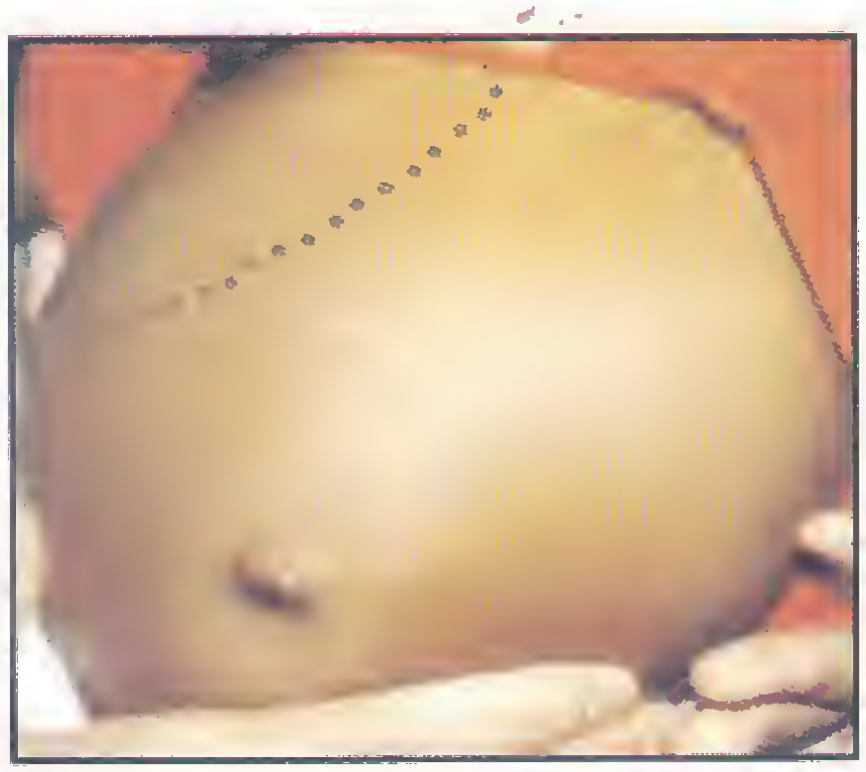
Phototherapy

When jaundice is more severe and bilirubin level is near to critical value (i.e. above 15 mg/dl), phototherapy for few or several days is useful in lowering bilirubin level. Note the eye cover to protect the eyes.

Neonatal cholestasis

It is a condition characterized by persistent jaundice, hepatomegaly and pale clay colored stool.

Neonatal hepatitis and extrahepatic biliary atresia are the 2 main causes. Early differentiation between these 2 conditions (by Hida scan and liver biopsy) is important because in biliary atresia, early surgical correction is indicated (Kasai operation).



3. Common Skin Lesions

Milia

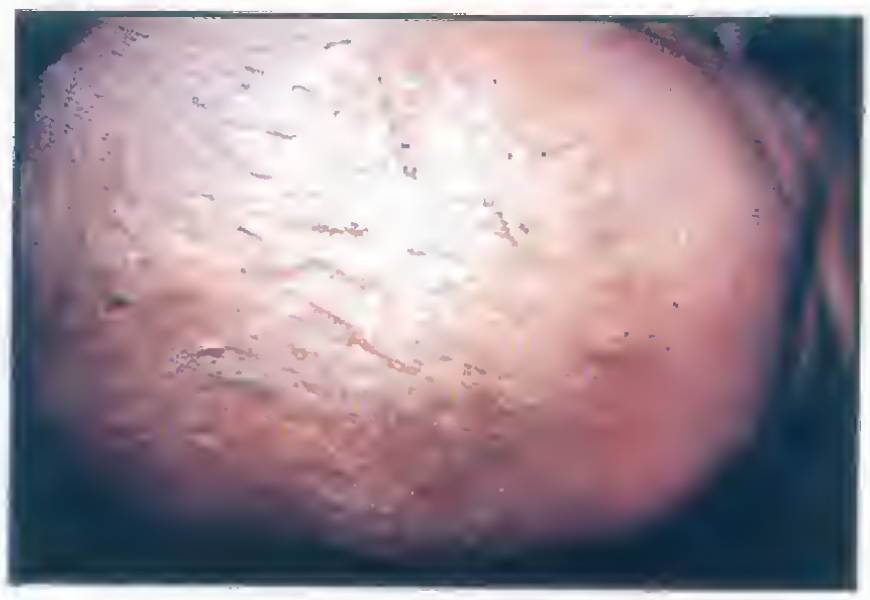
They are tiny sebaceous retention cysts scattered over the face, mainly on the nose and around it. They appear as whitish opalescent pinhead-sized spots that can be felt with the finger. It usually disappears within few weeks and no treatment is required

Infantile eczema

It is a form of atopic dermatitis that mostly starts in infancy. It mainly involves the face (cheeks) but scalp and extensor surface of extremities may be also affected. The lesions are intensely pruritic erythematous papules with scaling and roughening of the skin. It responds to topical steroids but the lesion has a recurrent nature.

Cradle cap

It is a form of seborrheic dermatitis that may start during the neonatal period. The scalp is the mainly involved site where diffuse or focal scaling and crusting occur (cradle cap). The lesions may involve the face, retroauricular area and diaper area. The condition responds to topical Steroids.



Common Skin Lesions

Erythema toxicum

It is a very common lesion that occurs in 50% of newborns during the first few days after birth. The lesions are firm yellow white papules or pustules (1-2 mm) with surrounding erythematous flare. The condition is benign and usually disappears over few days. The cause is unknown.

Diaper (Napkin) dermatitis

Erythematous diaper area due to prolonged contact with urine and stool. The lesion involves mainly the convex surfaces of buttocks, medial side of thighs and genital area but groin creases are usually spared. Secondary fungal infection (monilia) is common (see also minilial napkin dermatitis).

Mongolian spots

These are blue macular lesions that occur most commonly in the presacral area. They may be solitary or multiple and usually involve large areas. These lesions usually fade during the first 2 years.

4. Common Superficial Infections

Bullous impetigo

It is a common superficial infection in infants and young children caused staphylococcus aureus. It causes flaccid transparent bullae that mainly occur in diaper area and trunk. The bullae can easily rupture leaving a narrow rim at the edge of the moist lesion. Oral antibiotic therapy may be needed in extensive lesions.

Oral moniliasis

It is an acute infection of oral cavity caused by candida albicans. It is most common in neonatal period and early infancy. It is characterized by white flaky plaques covering all or part of the tongue, gingiva and oral mucosa. These plaques when removed leave a bright inflamed base. The condition usually subsides within one week of effective therapy with oral nystatin or miconazole.

Monilial napkin dermatitis

It usually follows contact dermatitis. It is characterized by intense fiery red erythema with sharp edge, which may extend to involve the whole napkin area and rises on the trunk. Small satellite lesions outside the sharp edge are characteristic of monilial napkin dermatitis. Treatment is by local antifungal Creams.

5. Common Umbilical Lesions

Umbilical sepsis (omphalitis)

The necrotic tissue of umbilical cord is an excellent medium for bacterial growth. Omphalitis may remain localized or may spread causing abdominal wall cellulitis (as opposite figure) and bacteremia. Portal vein phlebitis may occur and lead later to extrahepatic portal hypertension. Antibiotic therapy effective against Staph. aureus and E.coli is important

Umbilical granuloma

It is a moist granulation tissue at the base of the cord. The granuloma is soft, vascular and pinkish in color with mucoid or mucopurulent discharge. Cleaning with alcohol several times a day may be effective. Cauterization with silver nitrate (2 %) may be necessary in persistent cases.

Umbilical hernia

Umbilical hernia is a common anomaly that appear as a soft swelling covered by skin. It protrudes during crying, coughing or straining and it can be reduced easily through the fibrous ring of the umbilicus. Obstruction is unusual because of the wide ring. Most hernias that appear in the first 6 months disappear spontaneously before the age of one year.

6. Vascular Disorders

Salmon patch

Salmon patches (nevus simplex) are small ill-defined pink vascular macules that occur in 30-40% of normal newborn infants. The most common sites are glabella, eyelids, and back of the neck. Most lesions persist for several months but eventually fade and disappear.

Port-wine stain

Port-wine stains (nevus flammeus) are large sharply circumscribed pink to purple macular lesions. The head and neck are the most common involved sites. Most lesions are unilateral and usually end along the midline. Unlike salmon patches, port-wine stains are permanent lesions that do not disappear with time but in contrary they may become darker with time. When port-wine stain is localized to the trigeminal area of the face, especially around the eye, the diagnosis of "Sturge Weber syndrome" should be considered. However, it is important to note that port-wine stains are commoner as isolated lesions.

Hemangioma of the back of the neck and trunk

Hemangiomas are sharply demarcated bright red lesions that are protuberant and compressible. They may be present at birth or appear during the first two months. They may occur at any area of the body and may be small or large and single or multiple.

Multiple hemangiomas of the face

Most hemangiomas undergo a phase of rapid expansion, followed by stationary period and finally by spontaneous involution. Most lesions involute completely before the age of nine years. Treatment is not required and reassurance of parents is important.

Large lobulated hemangioma of the face

Large hemangiomas may interfere with vision or feeding and may lead to ulceration and bleeding. Treatment includes dilute bicarbonate soaks, gelfoam and compression therapy. Oral corticoteroid therapy for 4 weeks is effective in some patients.

7. Congenital Anomalies

Cleft lip and palate

The incidence of cleft lip with or without cleft palate is about 1 in 750 births. It can be just a small notch on the upper lip or it may cause complete separation and extension into the floor of the nose. It can be also unilateral (to one side) or bilateral. Surgical closure of cleft lip is made at the age of 3 months while closure of the palate is made before the age of 1 year to enhance normal speech.

Meningomyelocele

It is a fluctuant midline mass mostly in the lumbosacral region. It is a saclike cystic structure covered by a thin later or epithelialized tissue (meningocele is covered by normal skin). It causes neurological manifestations as bowel and bladder incontinence and may be flaccid paralysis of lower extremities (these problems are usually absent with meningocele). Surgical repair can be made in the first week but bowel and bladder incontinence persist.

Ambiguous genitalia

It is a genitalia in which sex cannot be identified. It is either due to virilized female (labial fusion and clitoral hypertrophy) or undervirilized male (bifid scrotum and micropenis). It is a medical emergency and birth certificate should not be filled before identification of the sex (see endocrinal disorders).

Talipes equinovarus

It is a common malformation that occurs due to intrauterine molding and fetal crowding. The foot is turned downwards and inwards so that the sole is directed medially. Orthopedic treatment should be started as early as possible in the first few days after birth.

Congenital hip dislocation

It is another common malformation that occurs due to intrauterine molding and fetal crowding. Limitation of abduction is the cardinal sign of hip dislocation. Examination for hip dislocation is made by *"Ortolani maneuver"*. With the hips and knees flexed at right angles, the thighs are abducted; a dislocated femoral head will clunk back into the acetabulum. This palpable clunk can be felt by the middle finger of each hand placed over each great trochanter. Both hips should be examined independently. Suspicion is confirmed by an x-ray or ultrasound. Orthopedic treatment should be started as early as possible.

Congenital Anomalies

Undescended testes

It is the common disorder that occurs in 4% of boys at birth. In 90% of cases, the testes can be palpated in the inguinal canal and in 10% of cases it is bilateral. In the majority of cases, the testis descends spontaneously during the first 6 months. If not, it will remain undescended and needs operation at 1 year old (orchipexy).

Congenital hydrocele

It is the common disorder that occurs in 1-2 % of boys at birth. It is an accumulation of fluid in the tunica vaginalis. It is a smooth and nontender swelling and transillumiation of the scrotum confirms the fluid-filled nature of the mass. Most cases resolve at the age of 1 year.

Inguinal hernia

It is the common disorder that occur in 4 % of newborns and it is 6 times commoner in boys. It can be unilateral or bilateral and it appears as a bulge in the inguinal region. The bulge is more visible during crying or coughing. The hernia may descent to the scrotal sac but it can be reduced by gentle pressure. Once diagnosed, surgical repair is indicated.

8. Birth Injuries

Caput succedaneum

It is a diffuse edematous swelling of the soft tissue of the scalp over the presenting part of the head. It may extend over the middle line and may be associated with ecchymotic patches. The edema subsides spontaneously within the first dew days.

Cephalohematoma

It is a subperiosteal hemorrhage that presents as a firm swelling limited to the surface of one cranial bone usually the parietal. The swelling does not appear except after several hours after birth. Anemia and jaundice are the main complications and phototherapy may be needed. It usually subsides gradually over 4-6 weeks.

Erb's palsy

Injury of brachial plexus is common and it occurs following traction on the head and neck. The affected arm is flaccid with forearm pronation and wrist flexion (waiter's tip position).

9. Abnormal Birth Weight

Premature baby

Low birth weight (below 2.5 Kg) is due to either premature delivery or intrauterine growth retardation (50% of cases for each). Premature baby has incomplete flexion of limbs and signs of prematurity are evident (incomplete sole creases, underdeveloped genitalia, small breast nodule, underdeveloped ear lobule and fine wooly scalp hair).

Intrauterine Growth Retardation (IUGR)

Fetal malnutrition due to placental insufficiency leads to IUGR and low birth weight. The head is relatively large and features of prematurity are absent. The skin is pale and looks dry with cracking and peeling.

Infant of diabetic mother

The baby is overweight, large, plump and plethoric. The face resemble that of patients receiving steroids. Hypoglycemia and hypocalcaemia are common and the baby tends to be tremulous and hyperexcitable. Cardiomegaly occurs in 30% of cases. The incidence of congenital anomalies is 3 times higher. Also the subsequent incidence of diabetes is higher than that of general population.

10. Chromosomal Syndromes (Trisomy 21)

Mongoloid features

Trisomy 21 (Down syndrome) is the most common autosomal trisomy (1/700 live birth). Recognition at birth is not difficult due to its characteristic features (upward slanting palpebral fissure, epicanthal folds, flat nasal bridge, simian crease and big space between first and second toes). Associated congenital heart disease occurs in 40% of cases. Delayed motor and mental development appears in infancy. In absence of congenital heart disease, long term survival is usual.

Simian crease and clinodactyly

Simian crease (single transverse palmar crease) is present in only 50% of cases. Clinodactyly (incurved little finger) is commonly present. It is important to note that simian crease is present in 4% of normal individuals.

Big space between first and second toes

Big space between first and second toes is one of the most constant findings in Down syndrome (in 97% of cases). Also upward slanting palpebral fissure is present in 97% of cases.

Chromosomal Syndromes (Trisomy 18)

Characteristic features

Trisomy 18 (Edward syndrome) is the second most common autosomal trisomy (1/4000 live birth). The main characteristic features are low birth weight, microcephaly, micrognathia (receding mandible), low set malformed ears, prominent occiput, clenched fist, syndactyly and rokerbottom feet.

Clinched fist

Clinched fist, syndactyly (fused digits or toes) and rokerbottom feet are commonly present limb anomalies. However, these anomalies are not peculiar to trisomy 18 and may be present in other morphological syndromes.

Associated congenital heart disease occurs in 60% of cases. Severe CNS malformations as hydranencephaly are common. Prognosis is poor as 30% of cases die in neonatal period and 90% die in infancy.

Syndactyly

Rockerbottom feet

Chromosomal Syndromes (Trisomy 13)

Coarse features

Trisomy 13 (Patau syndrome) is the third most common autosomal trisomy (1/6000 live birth). The main characteristic features are low birth weight, microcephaly and coarse features (low anterior hair line, microphalmia, micrognathia, hypotelorism, median cleft lip and palate and low set malformed ears). Associated congenital heart disease occurs in 80% of cases. Severe CNS malformations are common. Prognosis is poor as 50% of cases die in neonatal period and 90% die in infancy

Low set malformed ears

Low set malformed ears is usually present but this finding is common with many other chromosomal diseases.

Polydactyly

Polydactyly (extradigit) is common in trisomy 13 but again this finding is not peculiar and present in many other syndromes.

Chromosomal Syndromes (Turner Syndrome)

Webbed neck and low posterior hair line

Turner syndrome (45, X female) is a common chromosomal disorder of females (1/10.000) . It can be recognized at birth by the presence of webbed neck, low posterior hair line, widely separated nipples and edema of the dorsum of hands and feet. Later in childhood, it presents with short stature and gonadal dysgenesis (primary amenorrhea).

Widely separated nipples

Associated renal anomalies (40% of cases) and cardiac anomalies especially coarctation of aorta (20% of cases) may be present.

Edema of hands and feet

2

NUTRITIONAL DISORDERS

1. Marasmus.
2. Kwashiorkor.
3. Nutritional Rickets.

1. Marasmus

First degree marasmus
(loss of subcutaneous fat over the abdominal wall)

Marasmus is a severe form of undernutrition. It is characterized by weight loss, subcutaneous fat loss and muscle wasting. According to the degree of wasting, marasmus is is divided into 3 clinical grades:

First degree: Loss of subcutaneous fat over the abdominal wall.
Second degree: Loss of subcutaneous fat over the buttocks and thighs.
Third degree: Loss of subcutaneous fat over the face (senile or monkey face).

Second degree marasmus
(loss of subcutaneous fat over the buttocks and thighs)

Third degree marasmus
(Senile or monkey face)

2. Kwashiorkor

facial edema, hair changes and vitamin deficiencies

The kwashiorkor patient looks apathetic and miserable. Note the edema of the face, hair changes (the hair is lighter in color, sparse and easily detached) and angular stomatitis (ariboflavinosis or vitamin B_2 deficiency).

Kwashiorkor is a severe form of malnutrition caused by severe protein deficiency with excess carbohydrate intake. It occurs mainly between 6 months and 2 years (age of weaning).

The ***constant features*** of kwashiorkor are edema (face, dorsum of hands and feet), mental changes (apathy and anorexia), decreased muscle fat ratio and growth failure.

Variable features are hair changes, skin changes, anemia, vitamin deficiencies and hepatomegaly.

Skin changes

Skin changes occur mainly in the lower limbs, buttocks and napkin area where darkening, fissuring, scaling and hypopigmentation occur.

Edema of the dorsum of feet

3. Nutritional Rickets

Large head and frontal bossing

Most cases of nutritional rickets are seen between 6 months and 2 years. Skeletal changes of rickets involve the head, thorax, limbs and spine.

a) Head: Large head, frontal bossing, delayed closure of the fontanel and delayed teething.

b) Thorax: Rosary beads (enlarged costochondral junctions), longitudinal sulcus (vertical groove behind the rosary beads) and Harrison sulcus (horizontal groove at the lower costal margin). Chest deformities as flaring of lower ribs and pigeon chest (sternal protrusion) are seen in advanced cases.

c) Limbs: Broad epiphyses at wrist and ankle joints and Marfan sign (transverse groove palpated over the medial malleoli) are the main findings. Limb deformities as genu varum (bow legs) or genu vulgum (knock knees) are seen in advanced cases.

d) Spine: Correctable rounded kyphosis may occur.

Rosary beads

Broad wrists

Pigeon chest

Broad ankles

Bow legs

Knock knees

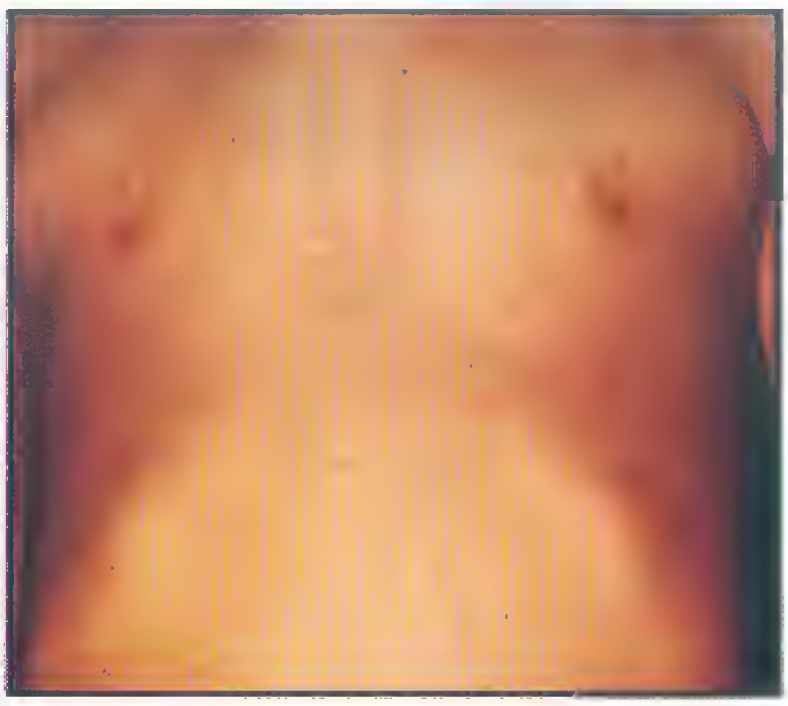
3

INFECTIONS

1. Scarlet fever.
2. Measles.
3. German measles.
4. Roseola infantum.
5. Chickenpox.
6. Herpes simplex.
7. Herpes zoster.
8. Superficial bacterial infections.
9. Superficial fungal infections.
10. Mumps.

1. Scarlet Fever

Fine papular rash (sandpaper appearance)



Scarlet fever is a bacterial disease caused by group A beta-hemolytic streptococci. The disease starts acutely with fever, vomiting and abdominal pain. The rash appears on the first or second day and soon becomes generalized as a red punctate or finely papular rash (goose-flesh or sandpaper appearance). The face appears flushed with circumoral pallor.

Strawberry tongue

The most important two features are the presence of sore throat and tongue changes. In early days, the tongue has a white coat with prominent papillae *"white strawberry tongue"*. After several days the white coat desquamates leaving a red tongue with prominent papillae *"red strawberry tongue"*

The rash remains for 3 - 7 days and fades with branny desquamation and peeling of finger tips.

Peeling of finger tips

2. Measles

Maculopapular rash on the face, trunk and limbs

Measles (Rubeola) is a viral disease. It starts with fever, which rises gradually during the first 4 days to reach 40.0°C with appearance of the rash. The fever is associated with severe catarrhal manifestations (rhinitis, conjunctivitis and cough).

The maculopapular rash appears on the 4th or 5th day. It starts in the face and within 2 days it covers the whole body. The rash characteristically reaches the feet while still in the face. It fades in the same order of distribution over the next 3 days (i.e. remains for 5-6 days).

The most important pathognomonic feature of measles is the *koplik's spots*. They appear as white grains of sand surrounded by red areola and located on oral mucosa opposite to the lower molar teeth. They appear on the third day of illness (one day before the onset of rash), remain for one or 2 days and disappear with appearance of the rash (i.e. it is an early diagnostic sign).

Koplik's spots

3. German Measles

Maculopapular rash on the face, trunk and limbs (remains for 2-3 days)

German measles (Rubella) is a viral disease. It starts with mild fever and lymphadenopathy. The fever remains only for 1-2 days and it may be absent *(cold measles)*.

The rash starts on the first or second day on the face and spreads rapidly to involve the trunk, then it extends rapidly to cover the whole body in 24 hours. The rash characteristically leaves the face while reaching the trunk and it clears completely by the third day *(3 day measles)*.

The most important characteristic feature is the *lymphadenitis* of the occipital, post-auricular and posterior cervical groups. Complications are almost absent in children.

Congenital rubella

German measles in pregnant women is serious to the developing fetus. Congenital rubella infection is associated with serious anomalies especially cataract and congenital heart disease.

Cataract of congenital rubella

4. Roseola Infantum

Rose maculopapular rash mainly on the trunk (remains only for 24 hours)

Roseola infantum (or Exanthem subitum) is a common viral disease of infants and young children.
The disease starts with a sudden fever, which rises rapidly to 39.5 - 41.0C°. It remains high for 3 - 4 days without any localizing signs then it falls by crisis with appearance of the rash.

The rash appears on the 4th day with the drop of temperature. It starts on the trunk and spreads rapidly to arms and neck with minimal face involvement. It fades very rapidly in 24 hours.
The rash can be considered as a good sign because complete recovery will occur over the next 24 hours. The most characteristic feature is the sudden drop of temperature with appearance of the rash.

Early diagnosis before appearance of the rash is difficult but the condition should be suspected in presence of high fever without localizing signs in late infancy. Febrile convulsions may occur with the sudden rise of temperature in susceptible infants.

5. Chickenpox

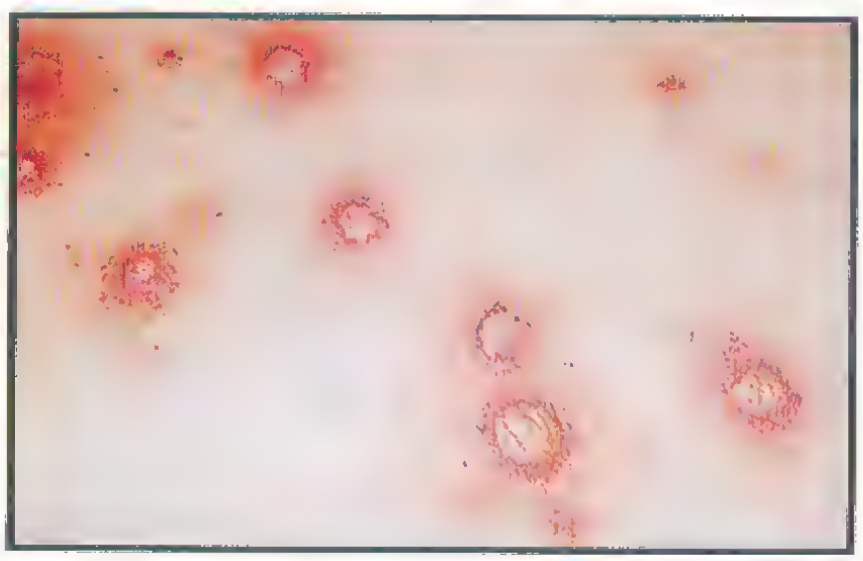
Pleomorphic vesicular rash

Chickenpox (Varicella) is a common viral disease that occur at any age including neonates. The disease starts with mild fever and the skin rash. The fever remains only for 1-2 days.

The vesicular rash appears first on the trunk and spreads to face and proximal parts of extremities (*centripetal distribution).* It appears in successive crops over 3 - 4 days. Each crop starts as a maculopapular, which rapidly vesiculates. Vesicles are oval teardrops surrounded by a red erythema. On the second day they change to pustules then crusts. So, at the peak of the disease the rash consists of crusts (earliest crop), pustules (next crop), vesicles and papules (latest crop)... i.e. the rash is *pleomorphic.* It is characteristically associated with itching *(pruritic).*

The whole duration of illness from the papules of first crop to the crusts of last crop is one week.

Tear drop vesicles with surrounding erythema



Chickenpox

Severe chickenpox with vesicles on oral cavity

Vesicular rash on the face and oral cavity (tongue, gingiva and hard palate).

Although chickenpox runs a benign course in most cases, serious and even fatal complications may occur especially in young infants and immunologically deficient patients. Secondary bacterial infection of the skin lesions may occur due to scratching. Hematological complications as thrombocytopenia and purpura fulminans may also occur. Neurological complications include encephalitis, Guillain Barre syndrome, facial nerve palsy and optic neuritis.

Neonatal chickenpox

Chickenpox in neonates and young Infants is very serious and may be even fatal. The disease is usually severe and complications are common. Thrombocytopenia and purpura fulminans (as opposite figure) are particularly very serious and may lead to fatal hemorrhage.

6. Herpes Simplex

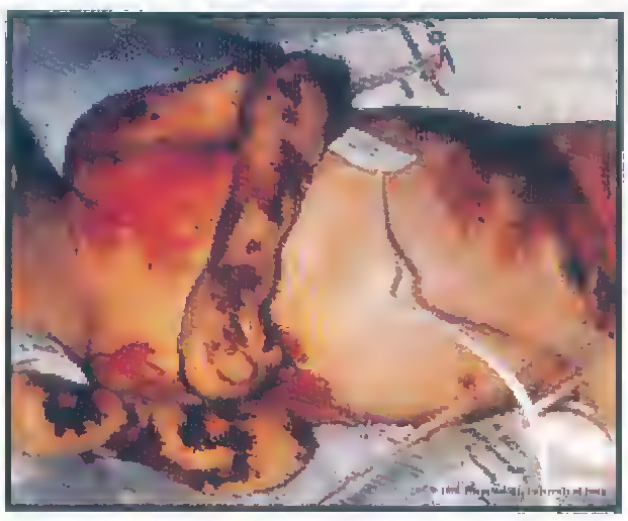
Uniform vesicular rash

Herpex simplex is a viral disease that may occur at any age including neonates.It disease starts with fever, which may reach 40.0°C or more and may remain as long as 10 days.

The rash appears on the second or third day. It mainly affects the *muco-cutaneous junctions* (the angles of the mouth or near genitalia) but it may be also *generalized*. The rash starts as vesicles , which are characteristically painful. They change to pustules and crusts over a period of 10 days. The rash is uniform and not pleomrphic as in chickenpox.

Neonatal herpes simplex

Neonatal herpes simplex is a serious life-threatening disease. Infection can be acquired during delivery or after birth. The disease can be disseminated with severe skin lesions as purpura fulminans. Mortality rate is high but therapy with intravenous acyclovir reduces the risk of death.



7. Herpes Zoster

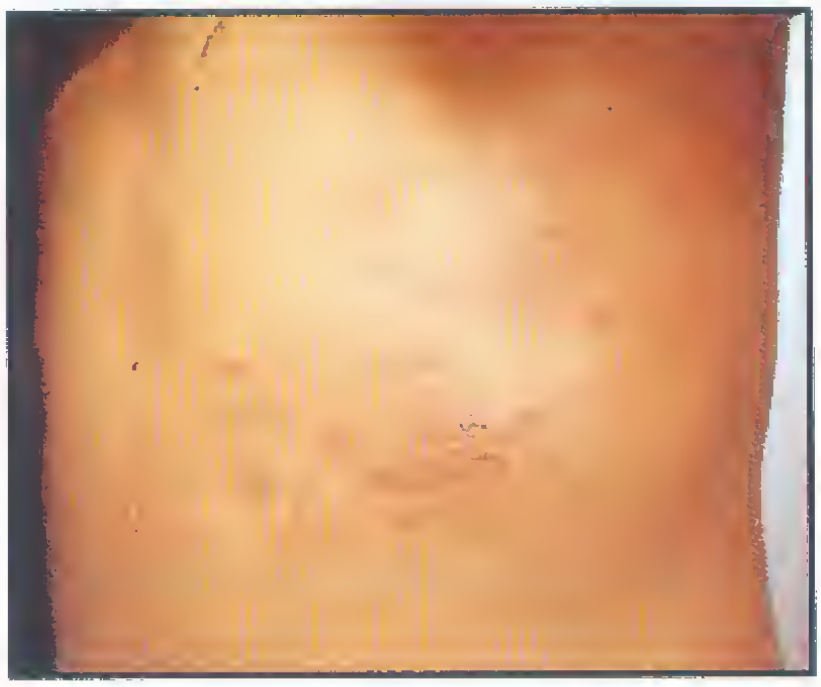
Unilateral vesicular rash confined to one dermatome

Herpes zoster (Shingles) is a viral disease that mostly occurs in adults. It is uncommon below the age of 10 years.

The disease starts with fever and pain along the involved dermatome. Fever may remain along the course of illness (2 -3 weeks).

The vesicular rash appears after few days and is characteristically *unilateral* and confined to a dermatome or 2 adjacent ones. The most commonly involved areas are the trunk but face, scalp and limbs may be affected. It starts as groups of palpules, which rapidly vesiculate, then become pustular and dry over a period of 5-10 days. Successive crops appear for 1-4 days along the course of the nerve. The rash remains for 2 -3 weeks. It is characteristically accompanied with *pain and tenderness* along the involved dermatome. The unilateral distribution of the rash and the accompanied pain and tenderness are the characteristic features.

In children, complications are unusual and the course is usually mild and prognosis is generally good. Transient paralysis of the affected part is rare. Other complications as post-herpetic pain and keratitis are rare in children. Treatment is by oral acyclovir



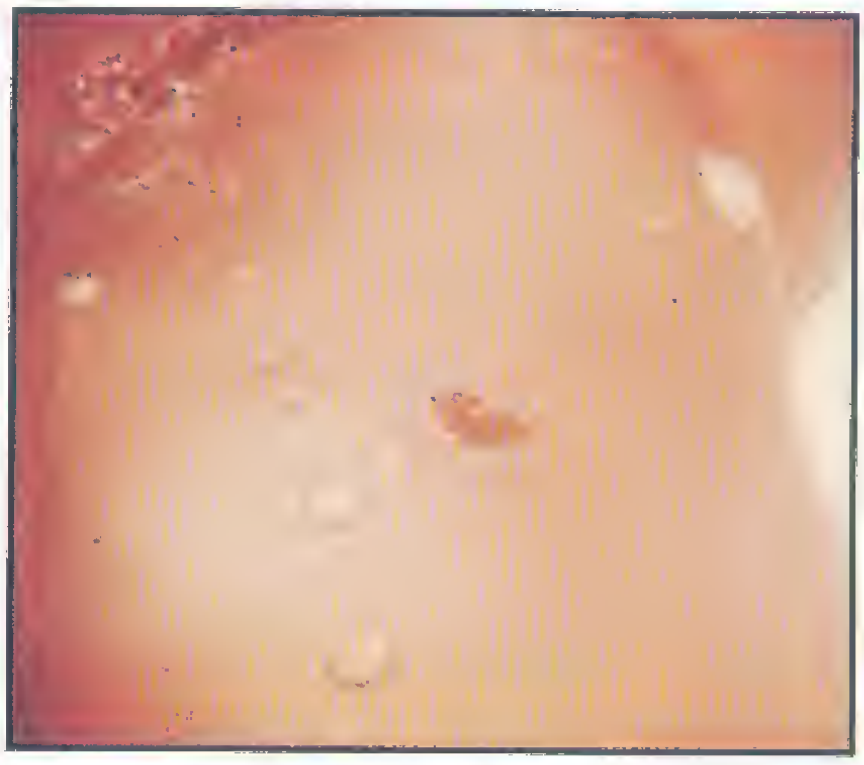
8. Superficial Bacterial Infections

Impetigo

It is a common skin infection caused by group A streptococci and occurs mainly in children during the hot summer season. The main sites of involvement are the exposed areas as the face, neck and limbs. It starts as erythematous macules that rapidly develop into thin-walled vesicles and pustules, which rapidly develop into sticky raised crusts. Treatment is by oral erythromycin or first generation cephalosporins.

Bullous impetigo

It is mainly an infection of infants and young children and always caused by staphylococcus aureus. Flaccid transparent bullae develop most commonly on the skin of the face, trunk, buttocks or extremities. Rupture of bullae occurs easily leaving a narrow rim of scale at the edge of the shallow erosion. Treatment is by oral erythromycin or first generation cephalosporins.



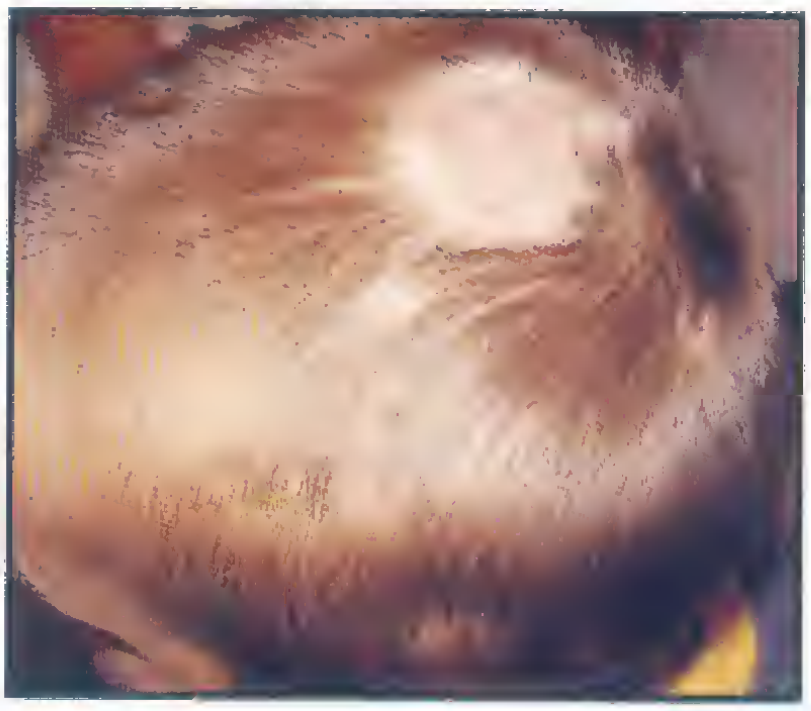
Scalded skin syndrome

It is a staphylococcal disease of infants and young children and it is considered as a severe form of bullous impetigo. The disease starts with fever and skin erythema. The erythematous skin rapidly acquire wrinkled appearance and flaccid bullae develop. Then, large sheets of epidermis peels away leaving moist glistening denuded areas. Healing occurs without scarring in 10-14 days. Treatment is by parenteral antibiotics.

9. Superficial Fungal Infections

Tinea capitis

It is a common fungal infection of the scalp mostly caused by trichophyton tansurans. It causes eryhthematous scaly circular plaques (ring worm) with localized hair loss. Broken hair follicles may appear as black dots (black-dot ring worm). Treatment is by oral griseofulvin for 8-12 weeks.



Tinea corporis

It is a common fungal skin infection mostly caused by different trichophyton species. It causes the characteristic scaly eryhthematous annular plaques that spread peripherally and clear centrally, giving the characteristic annular pattern. Treatment is by topical application of antifungal creams (as miconazole) for 2-4 weeks

Tinea versicolor

It is a common fungal skin infection Mostly caused by yeast malassezia furfur. The characteristic scally lesions vary in color. They are reddish brown in whites and hypopigmented or hyperpigmented in blacks. The most commonly involved areas are the neck, upper chest, upper arm and back. The lesions commonly enlarge and merge to form confluent patches. Treatment is by topical application of antifungal creams.

10. Mumps

Parotid swelling (unilateral or bilateral)

Mumps (Epidemic parotitis) is a viral disease, caused by myxovirus parotitis. It is the most common cause of acute parotid swelling. The parotid swelling occupies the parotid region over the angle of the mandible. It is continuous above and below the border of mandible and it elevates the ear lobule and pushes it upwards and outwards. The illness starts acutely with fever and swelling of one parotid gland. Swelling of the other parotid gland follows after 1-2 days. The swelling reaches its peak within 2-3 days and subsides slowly over 3-7 days. The swelling is painful and tender and better seen than felt as the edema of the skin and soft tissues usually extends and masks the limits of the swelling. The parotid swelling may be accompanied with swelling of other salivary glands (sublingual and submandibular glands). In some cases, swelling of submandibular glands occurs without parotid swelling. Although the course is generally benign in most cases, serious complications may occur. *Meningoencephalitis* is the most common and it is manifested by vomiting, headache, irritability and neck rigidity. *Pancreatitis* may also occur and leads to acute abdominal pain and vomiting. *Orchitis* is rare before puberty. Diagnosis can be confirmed by the presence of elevated serum amylase level.

4

NONINFECTIOUS SKIN LESIONS

1. Urticaria.
2. Papular urticaria.
3. Atopic dermatitis.
4. Seborrheic dermatitis.
5. Erythema multiforme minor.
6. Erythema multiforme major.
7. Kawasaki disease.
8. SLE and dermatomyositis.
9. Sweat rash.

1. Urticaria

Urticarial rash on trunk and limbs

The urticaria rash may be localized or generalized and it consists of erythematous circumscribed raised skin lesions (wheals). The individual lesion resolves within two days but new ones may continue to appear singly or in crops. The lesions are usually itchy and itching may be intense.

Causes of urticaria are very numerous and include *ingestants* (certain foods as fish, banana, chocolates, nuts or certain drugs as penicillin, sulphonamides, aspirin), *injectants* (as injection of drugs as penicillin, serum or blood transfusion, and insect stings), *inhalants* (as inhalation of pollens, danders or mites), *contactants* (as skin contact with drugs or chemicals), and *physical exposure* to cold (cold urticaria) or sun (solar urticaria).

Treatment is by oral antihistamines and local calamine lotion.

Typical raised urticarial wheals

2. Papular Urticaria

Papular rash mainly on limbs

Papular urticaria is a very common condition in children, which represents a delayed hypersensitivity reaction to insect bites. Fleas, ants and mosquitoes are the main responsible insects. The papular lesions may appear in large numbers and usually involve the extensor surfaces of extremities. The trunk may be also involved but the face and scalp are spared. The individual papule is about 2 - 5 mm in diameter and is surrounded by an erythematous area. Some papules, but not all, may vesiculate. The lesions are itchy and usually persist for more than 2 weeks. Recurrences are common. The condition should not be confused with other papular lesions especially scabies. In scabies the intensely pruritic eruption involves the interdigital spaces wrists, elbows, ankles, buttocks, umbilicus, groin and genitalia, and the papules are usually smaller in size than those of papular urticaria. Treatment is by oral antihistamines, local calamine lotion and avoidance of insect bites.

3. Atopic Dermatitis (Atopic Eczema)

Infantile eczema

It is a form of atopic dermatitis that mostly starts in infancy. It mainly involves the face (cheeks) but scalp and extensor surface of extremities may be also affected. The lesions are intensely *pruritic erythematous papules* with scaling and roughening of the skin. It responds to topical steroids but the lesion has a recurrent nature.

Infantile eczema

Several factors can increase pruritis and scratching as foods, inhalant allergens, bacterial infections, reduced humidity, excessive sweating and irritants as wool, soaps and detergents.

Atopic dermatitis is frequently associated with elevated level of IgE. 80% of patients develop allergic rhinitis or asthma.

Eczema herpeticum

Scratching of eczematous lesions can lead to secondary infection with herpes simplex (eczema herpeticum). Vesicles appear on eczematuos lesions and eventually turns to scabs. Recurrent attacks are common on chronic atopic lesions.

4. Seborrheic Dermatitis

Cradle cap

Seborrheic dermatitis is a chronic inflammatory disease common in infants and young children and may start in the neonatal period. The scalp is the most common involved site where diffuse or focal scaling and crusting occur (cradle cap).

Seborrheic napkin rash

The inflammatory process may involve the face, neck, retro-auricular area and diaper area. Diffuse erythema with scaling and crusting occurs.

Leiner disease

Leiner disease is a generalized severe seborrheic dermatitis associated with chronic diarhhea and dysfunction of immune system. The condition may resemble psoriasis and can be only differentiated with difficulty.

5. Erythema Multiforme Minor

Multiple skin lesions (maculopapular, vesicular and urticarial)

Erythema multiforme is a hypersensitivity reaction to a variety of causes as drugs, infections or exposure to toxic substances. The disease occurs in 2 forms (minor and major)

Typical iris or target lesions

Erythema multiforme minor is characterized by ***skin involvement only.*** The skin lesions are *variable and multiple* and may be maculo-papular, vesicular or urticarial. The lesions appear in crops for up to 3 weeks and affect mainly the extensor surface of extremities and trunk with symmetrical distribution. Palms and soles may be also affected. The characteritic skin lesion *"iris or target lesion"* is formed of urticarial lesions where dusky centers are surrounded by darker rings.

Vesicular lesions on hands

The skin lesions are not itchy and heal with hypopigmentation or hyperpigmentation but without scarring.

6. Erythema Multiforme Major (Stevens Johnson)

Early erythematous macules

Erythema multiforme major (Stevens -Johnson syndrome) is a severe serious form characterized by ***involvement of the skin and mucous membranes.*** The illness starts abruptly with fever and erythematous macules involving the face, body, palms and soles. These macular lesions change into bullae involving the skin, lips, mouth and conjunctiva.

Bullous lesions on the skin and mouth

New lesions erupt for 1 - 4 weeks and healing occurs during the next 6 weeks. Oral lesions are painful and interfere with feeding. Ocular lesions (mucopurulent conjunctivitis) may result in serious complications. Secondary bacterial infection of the denuded skin may result in septicemia and death.

Bullous lesions on the palm

Involvement of the palms and soles with bullous lesions is characteristic.

7. Kawasaki Disease

Strawberry tongue and cervical lymphadenopathy

Kawasaki disease (mucocutaneous lymph node syndrome) is a disease of unknown etiology mainly affecting children below the age of 5 years. The illness starts abruptly with high fever, which remains for one to several weeks.

Maculopapular rash

The fever is associated with at least 4 of the following 5:

1. Oral lesions including dry erythematous fissured lips and strawberry tongue.
2. Skin rash, which may be maculopapular or erythema multiforme but not vesicular,
3. Cervical lymphadenopathy, which is nontender, may be large and unilateral or bilateral.
4. Edema, erythema and peeling of finger tips.
5. Bilateral conjunctival injection.

Coronary vasculitis occurs in 30% of cases.

Edema and erythema of the palms

Prognosis is generally good and most cases recover completely over several weeks.

8. Systemic Lupus and Dermatomyositis

Systemic lupus
(Malar butterfly rash)

Systemic lupus erythematosus (SLE) is a disease of multisystem involvement mainly affecting females above the age of 8 years. Diagnosis of SLE depends on the presence of 4 or more of 11 criteria (malar butterfly rash is one of them). For more details, see Pediatric clinical diagnosis.

Dermatomyositis
(Heliotrope eyelids)

Faint malar rash with violaceous discoloration of eyelids.

Dermatomyositis is a multisystem disease characterized by a slowly developing muscle lesion and skin lesions. *Myositis* is manifested by weak, tender and indurated muscles mostly of the proximal groups. The *skin* over the involved limbs is usually thickened, tight and may be adherent to the underlying structures. A *dusky erythema* may cover the upper trunk and extremities. The most characteristic skin rash is the heliotrope eyelids (see above).

Dermatomyositis
(Erythema on extensor surface of extremities)

9. Sweat Rash (Miliaria)

Miliaria rubra

Sweat rash (or miliaria rubra) is a common inflammatory disease of the skin caused by mechanical obstruction of sweat ducts. It is common in infants especially in hot weather (in summer season).

The rash involves mainly the neck, trunk and diaper area but the limbs and face may be also affected.

The lesion is usually *fine papular* with intense erythema. The condition responds dramatically to cooling (by regulation of environmental temperature and by removal of excessive clothing). A cool bath is often helpful in relieving pruritis. Topical agents are ineffective and may exacerbate the eruption.

Miliaria crystallina

This form of miliaria is most common in newborns and young infants. It is characterized by *pin-point clear vesicles* that may cover large area of body surface. The condition is differentiated from bullous impetigo by the superficiality of the vesicles, clarity of fluids and absence of inflammation.

5

NEUROLOGICAL DISORDERS

1. Small and large head.
2. Floppy infant.
3. Cerebral palsy.
4. Duchenne muscular dystrophy.
5. Neurofibromatosis.
6. Tuberous sclerosis.
7. Sturge Weber syndrome

1. Small and Large Head

Familial microcephaly

Small head with severe affection of frontal lobe resulting in backward sloping forehead (camel head).
It is associated with severe mental retardation.

Congenital hydrocephalus

Neglected congenital hydrocephalus with large head, frontal bossing, dilated scalp and forehead veins and downward deviation of the eyes (setting sun eye sign). Hydrocephalus should be suspected and diagnosed earlier than that by serial measurement of head circumference and CT scan of the head.

Hydranencephaly

Large head with marked frontal bossing and associated anomalies (low set ears, micrognathia (receding mandible) and prominent occiput. Chromosomal karyotype proved that the baby is having trisomy 18.
Transillumination of the head proved that the large head is due to severe CNS anomaly (hydranencephaly).

2. Floppy Infant

Floppy infant is an infant with severe persistent hypotonia present at birth or in early infancy. Diagnosis depends on the presence three signs (frog leg position, head lag and curved trunk on ventral suspension).

Frog leg position

In supine position, the limbs are abducted and slightly flexed simulating frog legs. This denotes hypotonia of limbs.

Head lag

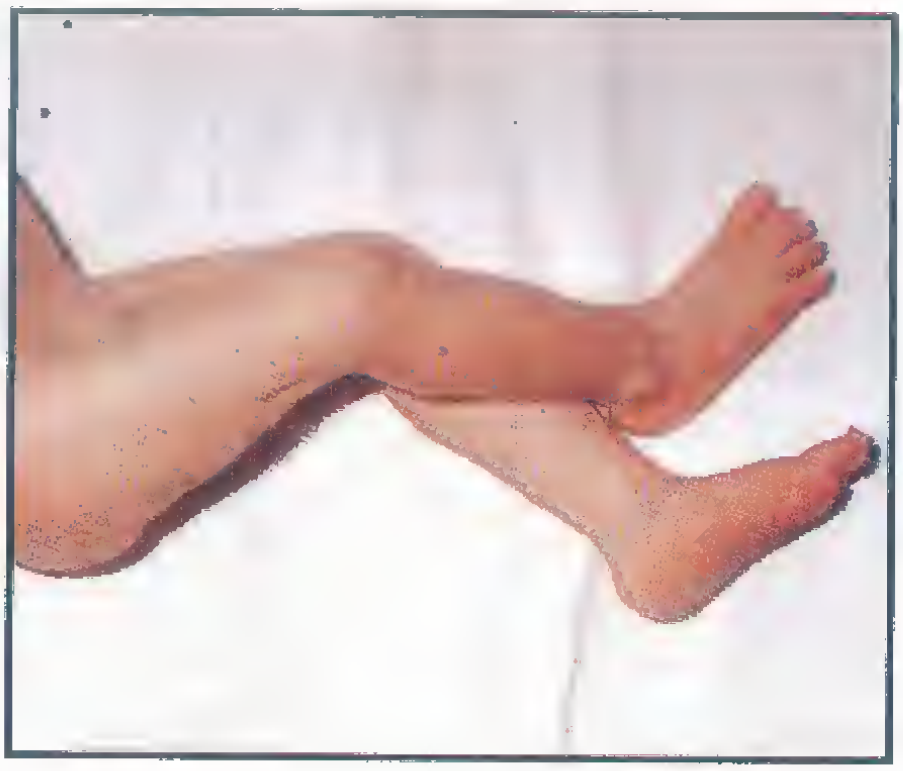
When the baby is pulled up from his hands, while in supine position, the head lags backwards. This denotes hypotonia of neck muscles.

Curved trunk on ventral suspension

When the baby is suspended in prone position over the examiner's palm, he droops around it. This denotes hypotonia of trunk muscles.

Werdnig-Hoffmann disease is the commonest cause of floppy infant. It is an autosomal recessive disease characterized by degeneration of anterior horn cells. Clinically, there is severe hypotonia, bulbar palsy and visible tongue fasciculations. Most cases die in the first 2 years by respiratory paralysis.

3. Cerebral Palsy



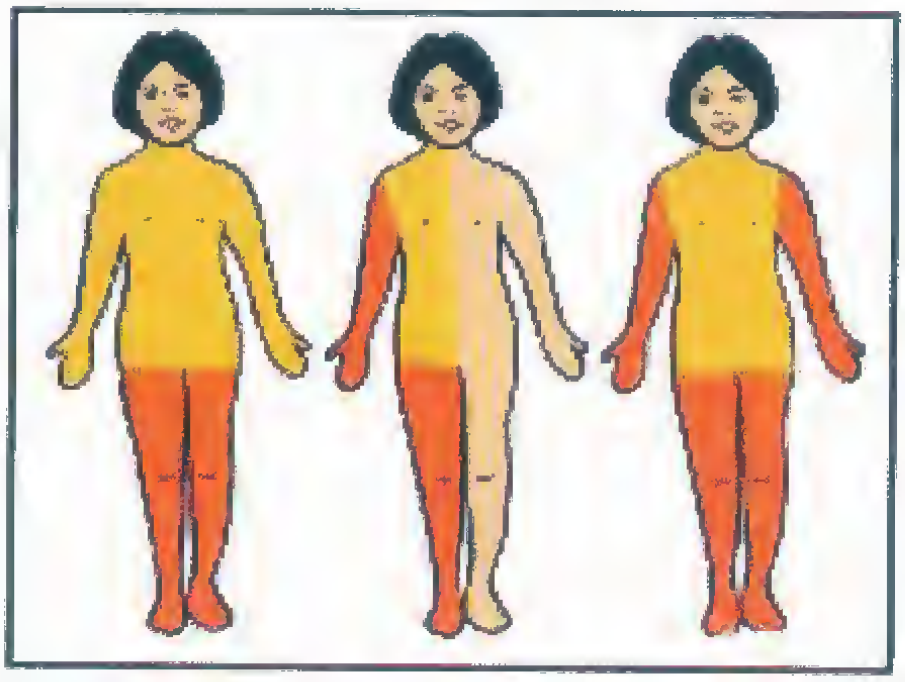
Spasticity and scissoring of lower limbs

Cerebral palsy is a term used to describe a heterogeneous group of disorders characterized by ***nonprogressive motor weakness*** resulting from a defect or lesion of the developing brain. According to the type of motor weakness, it is classified as atonic, spastic, extra-pyramidal and mixed.

Spastic cerebral palsy (the most common type) is characterized by hypoertonia and exaggerated tendon reflexes.

The distribution of spasticity is Variable. It may be *monoplegic* (only one limb is affected), *diplegic* (two limbs, usually the lower limbs), *hemiplegic* (one side is affected) and *quadriplegic* (four limbs are affected). With severe spasticity of lower limbs, scissoring of lower limbs occurs and ankle clonus becomes evident

Associated neurological signs as mental retardation, epilepsy, squint, pseudo-bulbar palsy and/or microcephaly may be present.



Diplegic **Hemiplegic** **Quadriplegic**

Physiotherapy

Physiotherapy is important to prevent contractures and muscle wasting.

CT scan of the head may show brain atrophy.

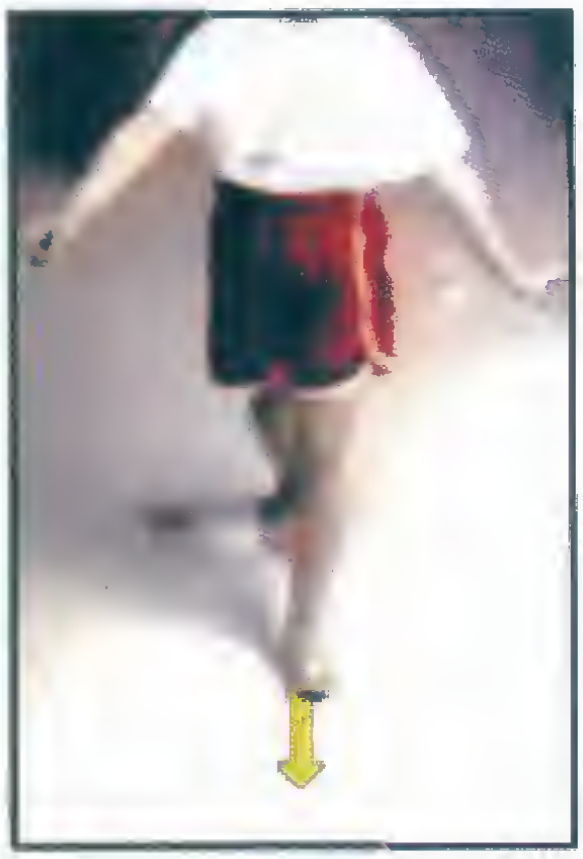
4. Duchenne Muscular Dystrophy

Duchenne muscular dystrophy is commonest cause of chronic ***progressive motor weakness*** in children. It is an x-linked disease appearing only in males with an onset of illness during the first 5 years and usually after infancy. The main characteristic features are:

a) Weakness of shoulder girdle muscles: The boy becomes unable to raise his arm above his head or to comb his hair. Slipping sign is positive (on trying to lift the child from the axillae, he slips through the examiner's hands).

b) Weakness of pelvic girdle muscles: The boy has a waddling unsteady gait. Difficulty in climbing stairs and in rising from the floor are evident. Gower's sign is positive (the boy rises from the floor by climbing up his legs).

c) Pseudohypertrophy of calf muscles: It is the most characteristic feature and it is usually associated with wasting of thigh muscles. Tongue and muscles of upper limbs may also show a pseudohypertrophy.



Waddling unsteady gait

d) Other features: Mild degree of mental retardation is usually present and cardiomyopathy is a constant feature and may be the cause of death.

e) The course is gradually progressive and most patients become unable to walk at the age of 12 years. *Death* usually occurs during the next 5 years due to respiratory failure or severe congestive heart failure.

f) Confirmatory investigations: Serum creatine phosphokinase is greatly elevated to thousands (normal level is below 60 units/liter). EMG shows nonspecific myopathic changes. Muscles biopsy is characteristic and diagnostic.

Gower's sign (the boy rises from the floor by climbing up his legs)

Note also the hypertrophied calf muscles (arrow).

5. Nurofibromatosis

Café-au-lait spots

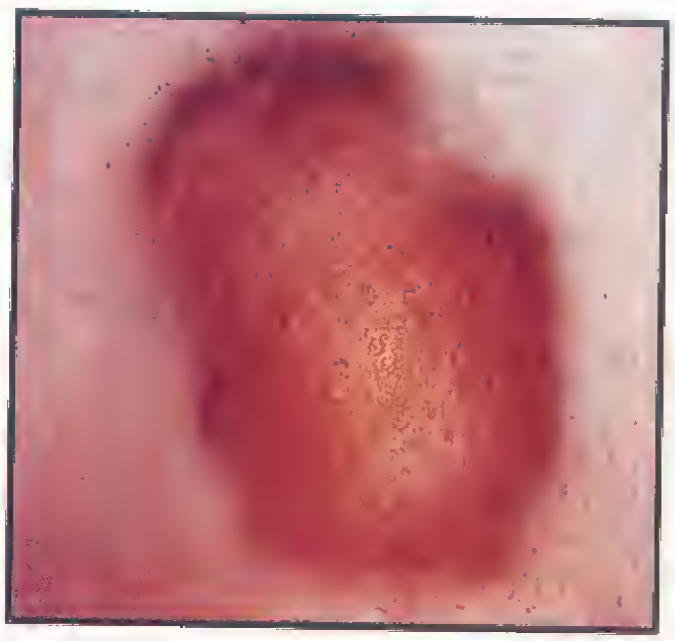
Neurofibromatosis is the commonest neurocutaneous syndrome (1/4000). It is an autosomal dominant disease characterized by skin and neurologic manifestations.

Cafe-au-lait spots are the most characteristic feature of the disease as they are present in 100% of cases. They are present at birth with tendency to increase in size, number and pigmentation during thefirst few years of life. Presence of more than 5 spots greater than 5 mm in diameter is diagnostic. *Neurofibromas* of the skin and subcutaneous tissues usually appear in late childhood. Plexiform neurofibroma is a large infiltrative tumor causing disfigurement.

Neurological manifestations include learning and speech difficulties.

Brain tumors (optic glioma, meningioma, neurofibroma or astrocytoma) are common and they are the principal risk and the main cause of death.

Plexiform neurofibromas



Neurofibromas

6. Tuberous Sclerosis

Hypopigmented macules

Tuberous sclerosis is the second most common neurocutaneous syndrome (1/30000). It is also an autosomal dominant disease characterized by skin and neurologic manifestations.

Hypopigmented macules on the trunk and limbs are present at birth in almost all cases. They are oval or irregular and ranging from few mm to several cms.

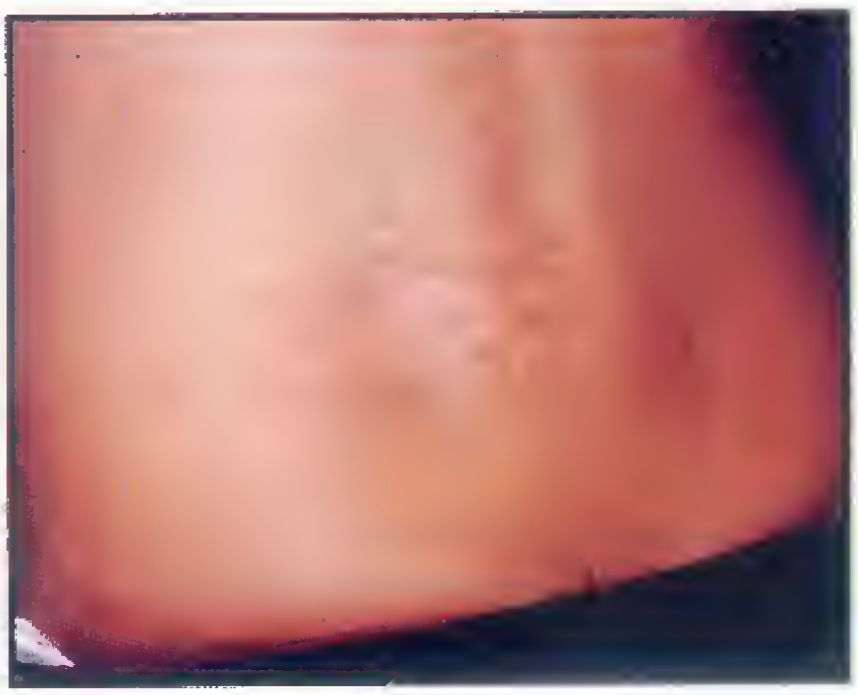
Sebaceous adenomas

Sebaceous adenomas are the most characteristic lesion that usually appears at 4 - 6 years. They are small bright red or brownish nodules on the nose and cheeks (butterfly distribution), which may be confused with acne.

Shagreen patches

Shagreen patches are a slightly raised indurated areas mainly located in lumbosacral region.

Neurological manifestations include epilepsy (in 90% of cases) and mental retardation (in 60% of cases). Behavioural disorders as hyperactivity and destructiveness are common.



7. Sturge Weber Syndrome

Unilateral port-wine nevus of the face

Sturge Weber syndrome is the third most common neurocutaneous syndrome (1/50000). It is a sporadic nongenetic disease.

Unilateral facial port-wine nevus is the most important feature of the disease. It usually involves the upper face and the eye lids but lower face may be also affected. However, not all children with facial nevus have Sturge Weber disease (see also port -wine stain in newborn chapter).

Buphthamos and glaucoma of the eye on same side of the lesion are common. Neurological manifestations include unilateral convulsions and hemiparesis on the other side of the lesion. Learning difficulties or mild mental retardation occur in late Childhood.

6

DIGESTIVE AND RENAL DISORDERS

1. Painful oral lesions.
2. Diarrheal dehydration.
3. Viral hepatitis.
4. Hepatosplenomegaly.
5. Veno-occlusive disease.
6. Nephrotic syndrome

1. Painful Oral Lesions

Monilial stomatitis

It is an acute infection of oral cavity caused by candida albicans. It is most common in neonatal period and early infancy. It is characterized by white flaky plaques covering all or part of the tongue, gingiva and oral mucosa. These plaques when removed leave a bright inflamed base. The condition usually subsides within one week of effective therapy with oral nystatin or miconazole.

Herpetic gingivostomatitis

It is an acute infection of the oral cavity caused by herpes simplex virus. It is most common between the ages of 1 - 3 years. Clinically, the condition starts with high fever, severe mouth pain, salivation and refusal of feeding. Examination of oral cavity reveals small ulcers (2-10 mm in diameter) over the tongue, gingiva and oral mucosa. In severe cases, ulcers may involve all oral mucosa.

Herpangina

It is an acute infection of oral cavity caused by coxsackievirus. It is mostly seen in children below the age of 5 yrs. Clinically, there is fever, sore throat and discrete vesicles and ulcers mainly located on the anterior tonsillar pillars. The ulcers are small in size and few in number (1-5). In severe cases, ulcers may involve the soft palate, uvula, tonsils and posterior pharyngeal wall.

Hand foot and mouth disease

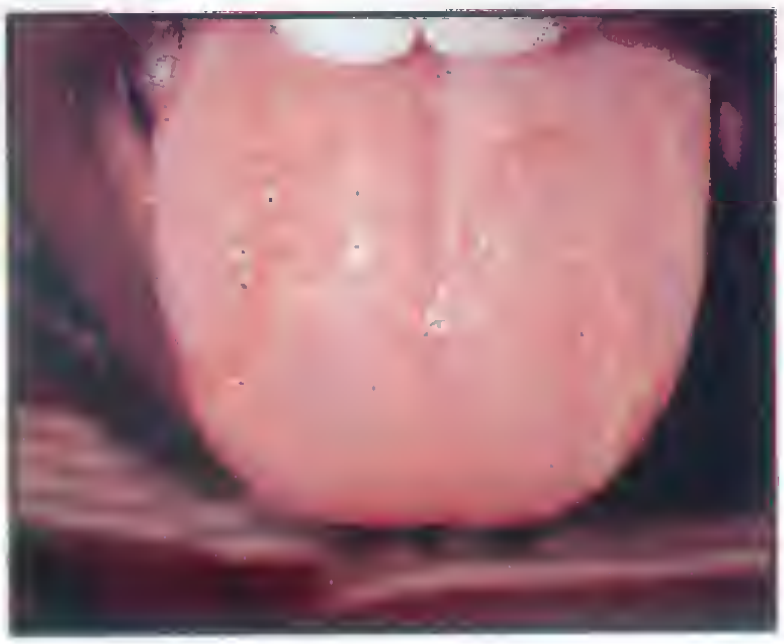
It is an another acute disease caused by coxsakievirus. It is characterized by vesicles and ulceration in the mouth and vesicles on the hands and feet.

Aphthous ulcers

It is a condition of unknown etiology with a tendency to recurrence. It is characterized by a solitary or few painful ulcers mainly in the floor of the mouth, ventral surface of the tongue and the mucobuccal fold. The ulcer is usually small (less than 5 mm) with a depressed center and erythematous periphery. Healing occurs in less than a week.

Geographic tongue

It is a common benign condition, which may be misdiagnosed as oral moniliasis or herpetic gingivostomatitis. It is characterized by the presence of one or more bright red patches with whitish or grayish margin upon the dorsum of an otherwise normally roughened tongue. The patches look like a map (geographic tongue). The condition is asymptomatic and may persist for several weeks or months. Treatment is not required.



2. Diarrheal Dehydration

Sunken eyes and dry tongue

Severe gastroenteritis is the most common cause of dehydration in infants and young children.

Clinical *signs of dehydration* are sunken eyes, dry tongue and lost skin turgor.

The *degree of dehydration* (mild, moderate or severe) depends on the severity of signs and the degree of weight loss.

Lost skin turgor

The *type of dehydration* (isonatremic, hyponatremic or hypernatremic) can be clinically suggested by the relative losses of water and electrolytes:

- In isonatremic dehydration, dryness of the tongue and lost skin elasticity are proportionate.
- In hypernatremic dehydration, the tongue is very dry and skin elasticity is not severely affected.
- In hyponatremic dehydration, the tongue is moist and skin elasticity is severely affected.
- Accurate differentiation is by measurement of serum sodium level.

Other *associated complications* should not be overlooked especially shock, metabolic acidosis, acute renal failure and electrolyte disorders as hypokalemia and hypocalcemia. Convulsions and bleeding (hypoprothrombinemia or DIC) may also occur.

Treatment of moderate to severe cases is by I.V fluid therapy over 24 hours. The treatment includes shock therapy, deficit therapy and maintenance therapy.

Dark urine (bilirubinuria)

Viral hepatitis (infection with hepatic viruses) is a common health problem. Clinical manifestations are divided into 3 stages (preicteric, icteric and convalescent stages).

a) Preicteric stage is characterized by mild fever, anorexia, vomiting and abdominal pain. This stage usually lasts for 4-6 days and the urine usually becomes dark (bilirubinuria) during the last 1- 3 days.

Jaundice

b) During the *icteric stage,* jaundice appears and the liver is enlarged and tender. The early manifestations as fever, vomiting and abdominal pain disappear but anorexia may continue. This stage usually lasts for 2- 4 wks.

c) Convalescent stage starts with complete resolution of hepatic injury. During this stage some children may continue to complain of malaise and fatigue for several weeks.

Tender Hepatomegaly

Laboratory diagnosis of acute hepatic injury includes direct or mixed hyperbilirubinemia and raised serum transferases.

Laboratory identification of the causative agent of acute hepatitis (A, B or C) depends on detection of hepatitis viruses markers.

4. Hepatosplenomegaly

In neonatal period and early infancy

In this age group, liver is usually bigger than the spleen because liver diseases are the main causes.

The main associated clinical finding is ***jaundice.***

The main causes are:

1. Neonatal hepatitis.
2. Extrahepatic biliary atresia.
3. Metabolic liver diseases as galactosemia and tyrosinemia.

In late infancy and early childhood

In this age group, spleen is usually bigger because liver diseases are not the main causes.

The main associated clinical finding is ***anemia.***

The main causes are:

1. Metabolic diseases (as Gaucher disease and Neimann Pick disease).
2. Chronic hemolytic anemia.
3. Acute leukemia.

In late childhood

In this age group, liver is usually bigger than the spleen because liver diseases are the main causes.

The main associated clinical finding is ***jaundice.***

The main causes are:

1. Chronic hepatitis.
2. Metabolic liver diseases as Wilson disease.
3. Bilharziasis in rural endemic areas.

5. Veno-occlusive Disease

Hepatomegaly, ascites and dilated abdominal wall veins

Veno-occlusive disease is an intrahepatic obstruction of hepatic veins by thrombotic lesions, which results in a *postsinusoidal portal hypertension*. The cause is unknown, but ingestion of herbal hepatotoxins may be responsible.

Clinical diagnosis depends on the presence of hepatomegaly, rapidly developing ascites and dilated abdominal wall veins in a malnourished child between the ages of 1- 4 years. Jaundice is usually mild or absent and spleen is not enlarged. Prognosis is generally bad as most cases deteriorate and die within several months of onset. Some cases may live longer and develop liver cirrhosis.

6. Nephrotic Syndrome

Facial edema (Puffy face and swollen eyelids)

It is the most common cause of generalized edema in children. It is a clinico-laboratory syndrome of 4 components:

1. *Generalized edema:* Swollen eyelids, puffy face and edematous limbs. Scrotal edema (in males), abdominal wall edema, and ascites occur in advanced cases.
2. *Massive proteinuria:* Above 2 gm/24 hours. Levels above 5 -10 gm/24 hours may occur.
3. *Hypoproteinemia:* Serum albumin below 2.5 gm/dl.
4. *Hypercholesterolemia:* Serum cholesterol level above 300 mg/dl.

Abdominal wall edema

Minimal change disease is the most common in children. It is characterized by:

1. Age of onset is between 2 - 6 years.
2. There is no significant or persistent hypertension or hematuria.
3. Proteinuria is selective (mainly albumin).
4. Excellent therapeutic response to corticosteroid therapy.
5. It is a disease of remissions and exaeerbations. Several relapses may occur but without deterioration of renal function.
6. Renal biopsy: Under light mieroscope, the glomeruli are free. Electron microscopy reveals fusion of the epithelial foot proeesses of the glomerular capillary wall.

Scrotal edema (in males)

7

HEMATOLOGIC DISORDERS

1. Beta thalassemia major.
2. Idiopathic thrombocytopenic purpua (ITP).
3. Disseminated intravascular coagulation (DIC).
4. Henoch-Schonlein purpura.
5. Hemophilia.

1. Beta Thalassemia Major (Cooley's Anemia)

Anemia (pallor) and Mongoloid features

Anemia, large head, prominent maxilla and mongoloid features.

The clinical triad of chronic anemia, mild jaundice and splenomegaly (or hepatosplenomegaly) is characteristic of chronic hemolytic anemia. Positive family history and history of repeated transfusions may be obtained. Growth failure (short staure) and skeletal changes (large head, prominent maxillae, mongoloid features) may be prominent especially in severe cases of Cooley's anemia.

Short stature and hepatosplenomegaly

Laboratory diagnosis of chronic hemolytic anemias depends on the presence of chronic anemia, sustained reticulocytosis above 2%, high serum iron (above 150 mcg/dl) and low iron binding capacity.

Important causes of chronic hemolytic anemias are hereditary spherocytosis, thalassemias and sickle cell anemia.

Hemoglobin electrophoresis is essential for diagnosis of hemoglobinopathies. In beta thalassemia major, high level of hemoglobin F (above 50%) is usually present.

2. Idiopathic Thrombocytopenic Purpura (ITP)

Ecchymotic patches on the abdomen

Purpura is a small hemorrhage into the superficial layers of the skin and mucous membranes that produces areas of purple discoloration, which do not blanch on pressure. Minute spots of 1-2 mm in diameter are known as *"petechiae or petechial spots"*.Larger areas of 1-2 cm in diameterare known as *"ecchymoses or ecchymotic* patches". both lesions are frequently present together.
In most cases, the skin of the trunk and limbs are the most commonly involved area. Bleeding is the main complication and it can be mild or severe. Bleeding gums and epistaxis are the most common while intracranial hemorrhage is the most serious.

Idiopathic thrombocytopenia purpura (ITP) is the most common cause of purpura in children. The illness is mostly immune in origin and it is often preceded by a viral infection 1 - 4 weeks before the onset of illness. The onset is abrupt with purpura and may be bleeding. Laboratory diagnosis depends on the presence of severe thrombocytopenia (platelet count is usually below 20.000). Prognosis is excellent in most cases. Purpura and bleeding usually subside over 1-2 weeks.
Persistence of purpura and thrombocytopenia for more than 2-3 weeks in an indication for bone marrow examination and lenkemia or aplastic anemia should be excluded.

Petechial spots

Ecchymotic patches on the legs

3. Disseminated Intravascular Coagulation (DIC)

DIC or disseminated intravascular coagulation is a serious disease characterized by consumption of platelets and some coagulation factors (1, 2, 5, 8) in a process of formation of minute intravascular clots. It occurs as a complication of another severe systemic illness. The main precipitating factors are septicemia, shock and acidosis. Gastroenteritis with dehydration, shock and acidosis is an ideal situation for DIC to develop.

Clinically, the patient is critically sick and the features of the precipitating disease are well evident (as septicemia, shock, acidosis). The hematological manifestations of the disease include bleeding from puncture sites and surgical incisions, purpura (petechiae and ecchymoses) and necrotic skin patches (characteristic of DIC). Internal hemorrhage (including intracranial hemorrhage) may occur due to thrombocytopenia and severe coagulation defect (see also chickenpox and herpes simplex).

Laboratory diagnosis depends on the presence of thrombocytopema, severe coagulation defect (prolonged thrombin, prothrombin and partial thromboplastin times) and Fibrin degradation products (FDPs) in the peripheral blood.

Prognosis is generally bad and it depends on the proper control of the precipitating factors and the extension of internal hemorrhage.

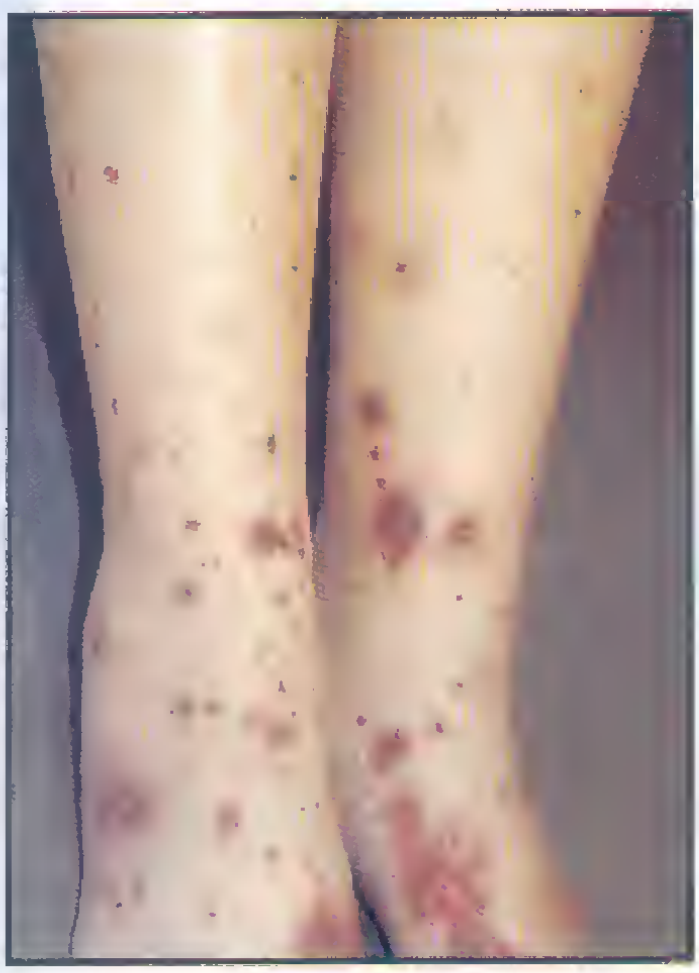
Ecchymotic patches

Necrotic skin patches

4. Henoch-Schoenlein Purpura

Purpuric rash on buttocks and back of lower limbs

Henoch-Schoenlein purpura is a vasculitis syndrome of nonthrombocytopenic purpura, arthritis, abdominal pain and nephritis. Characteristic skin lesion is a ***purpuric rash*** involving mainly the back of lower limbs and buttocks but it may extend to involve the trunk and upper limbs. ***Arthritis*** occurs in two thirds of cases, affecting few large joints and remains for only few days. ***Gastrointestinal manifestations*** occur in 50% of cases. Colicky abdominal pain is the main feature and it may be associated with gastrointestinal hemorrhage (bleeding per rectum and may be hematemesis). ***Nephritis*** occurs in one third of cases and it may appear during the acute stage or few weeks after recovery. Prognosis is generally excellent and most cases recover completely over few days or few weeks. There are no specific laboratory findings and the diagnosis is clinical.



5. Hemophilia

Bruising and hemarthrosis of the left elbow joint

Hemophilias are a group of inherited disorders that result from a defect in phase I coagulation. There are 4 types that differ in incidence, mode of inheritance and severity of bleeding.

Hemophilia A (classic hemophilia) It is the most common type (80% of cases). It is an x-linked disease caused by deficiency of factor 8 (antihemophilic factor). The clinical severity depends on the level of factor 8 activity in plasma. The most characteristic features of hemophilia A are spontaneous or traumatic hemorrhages which can be subcutaneous, intramuscular or within joints (hemarthrosis). In infants, excessive bleeding may follow circumcision, but bleeding is usually not evident in the first year of life. After infancy, with the time the child begins to walk, easy bruising and hemarthrosis become evident.

Laboratory diagnosis depends first on the presence of prolonged partial thromboplastin time (phase I defect).

Hemophilia B (Christmas disease) is the second most common type of hemophilia (about 10% of cases). It is an x-linked disease caused by factor 9 deficiency. Clinically, it cannot be differentiated from hemophilia A.

Intramuscular hematoma of the left calf muscle

Hemarthrosis of the right ankle joint

8

ENDOCRINAL DISORDERS

1. Ambiguous genitalia.
2. Hypothyroidism.
3. Short stature.
4. Precocious puberty.

1. Ambiguous Genitalia

Ambiguous genitalia is a genitalia in which the sex cannot be identified. It is a medical emergency requiring urgent and appropriate measures to identify the true sex. Parents can be told that the baby is either a *"female with overdeveloped genitalia"* or *"male with underdeveloped genitalia"*. Birth certificate should not be filled out until the sex is identified.

Ambiguous genitalia can result from one of 3 possibilities:

1. 46 XX-intersex (verilized female): The genitalia is ambiguous due to over-development by excess androgens (clitoral hypertrophy and labioscrotal fusion). Chromosomal analysis reveals XX and ultrasound genitogram reveals normal mullerian structures (vagina, cervix and uterus). The main causes are congenital adrenal hyperplasia and maternal virilization. Measurement of 17 ketosteroids in urine is useful in differentiation (elevated in congenital adrenal hyperplasia).

2. 46 XY-intersex (undervirilized male): The genitalia is ambiguous due to underdevelopment by androgen deficiency (small phallus and bifid scrotum). Chromosomal analysis reveals XY and ultrasound genitogram reveals absent or abnormal mullerian structure (vagina only without uterus). Hormonal studies and testicular biopsy are important to identify the responsible defect. The main causes are defects in testosterone synthesis or in testosterone action.

3. True gonadal intersex: The genitalia is ambiguous due to failure of differentiation. Chromosomal analysis (genotype) reveals XX, XY or mosaicism. Pelvic ultrasound usually reveals some mullerian structures. Diagnosis is established by gonadal biopsy, which demonstrates the presence or both gonads (ovary in one side and testis on the other side or bilateral ovotestes).

2. Hypothyroidism

Congenital hypothyroidism

Congenital hypothyroidism is mostly caused by aplasia or hypoplasia of the thyroid gland (90% of cases). Defective synthesis of thyroid hormone and iodine deficiency are uncommon causes. The condition should be diagnosed early in neonatal period or early infancy before the development of mental retardation and coarse features (large head, coarse hair, low anterior hair line, swollen eyelids, depressed nasal bridge and thick protruded tongue). It is not useful to diagnose a classic case of **cretinism** with coarse features and mental retardation (opposite figure).

Neonatal thyroid screening program is recently implemented in Egypt where all newborns are screened for hypothyroidism between the third and the 7th day of life. A blood drop is obtained by heel prick on a filter paper, and is analyzed for TSH and T4.
- If TSH value is > 20 micro unit/L with low or normal T4 value, an immediate blood sample is withdrawn and reanalyzed for T4 and TSH to confirm data obtained from filter paper and treatment is immediately initiated.

Treatment is by life long therapy with oral thyroid hormone. The dose is 10 microgram/ kg/ day at birth and in infancy.

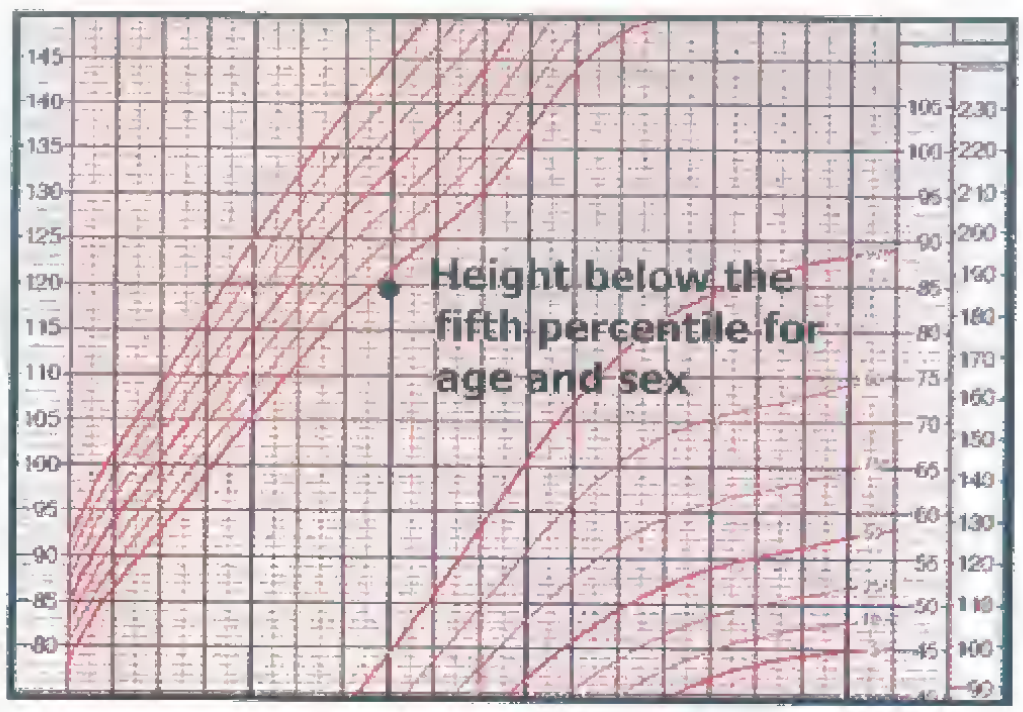
Thyroid enlargement (Goiter)

Acquired hypothyroidism

Acquired hypothyroidism is mostly Caused by Hashimoto thyroiditis, which it is an autoimmune disease (circulating thyroid antibodies can be detected in the blood).The main 3 clinical presentations of acquired hypothyroidism are short stature, goiter and school underachievement. Presence of any of these presentations is an indication for measurement of thyroid hormone

3. Short Stature

Short stature is a height below the fifth percentile for age and sex. Causes can be classified into 2 groups (disproportionate and proportionate).



1. Disproportionate short stature is caused by skeletal dysplasias causing short limbs as achondroplasia and osteogenesis imperfecta (see opposite page).

2. Proportionate short stature is caused by 3 main groups:

a) Normal variants: Genetic (familial) short stature and constitutional growth delay.

* In genetic (familial) short stature, one parent at least is short. Bone age is normal and puberty occurs at the usual age. The ultimate adult height is short or midway between the heights of both parents.

* In constitutional growth delay, the height of both parents is normal but history of slow growth in childhood in other family members may be obtained. Bone age is delayed and puberty will be also delayed.
Because of delayed bone age and puberty, the predicted ultimate adult height is normal.

b) Chronic systemic diseases: Chronic malnutrition, chronic cardiac disease, chronic respiratory disease or chronic renal failure.

c) Endocrinal diseases: Hypothyroidism, growth hormone deficiency and excess glucocorticoids (Cushing syndrome). Prolonged corticosteriod therapy gives features similar to Cushing syndrome with facial edema (moon face), hirsutism and growth failure.

Cushinoid face of prolonged corticosteroid therapy

Achondroplasia

Achndroplasia is an autosomal dominant disease characterized by clinical triad of short limbs, large head and normal trunk. The shortening of limbs is involving the proximal segments (the humerus and femur). Note also the forking of fingers and the exaggerated skin creases of limbs.

Osteogenesis imperfecta

It is an autosomal dominant disease characterized by excessive bone fragility, multiple fractures (healing with angulated deformities) and blue sclera. Note the blue sclera and angulated deformities of the right humerus and both lower limbs.

4. Precocious Puberty

True precocious puberty in male (enlarged penis and testes with pubic hair appearance and high gonadotropin level)

Precocious puberty is the early appearance of secondary sexual characters before 8 years in girls or 9 years in boys. Secondary sexual characters are breast enlargement in girls and pubic and axillary hair in boys and girls.

Clinical criteria for diagnosis are:

*** In males**

Enlarged testes (more than 2 cm in greatest length) before 9 years and/or enlarged penis (more than 7 cm in stretched length) before 10 years.

*** In females**

Breast enlargement before the age of 8 years, pubic or axillary hair before the age of 9 years and/or menarche before the age of 10 years.

a) In true precocious puberty, the gonadotropin level is high (gonadotropin dependant) and gonads are enlarged (testes in males, ovaries in females). Spermatogenesis occurs in males and ovulation occurs in females. True precocious puberty is always isosexual.

Precocious pseudopuberty in female (breast enlargement without ovulation and with low gonadotropin level)

b) In precocious pseudopuberty, the gonadotropin is low (gonadotropin independent) and gonads do not enlarge. Spermatogenesis or ovulation does not occur. It can be either isosexual or heterosexual.

c) In partial precocious puberty, only one isolated manifestation of puberty appears as breast enlargement (premature thelarche), pubic hair appearance (premature adrenarche) or menses (premature menarche). In all these conditions, other secondary sexual characters are absent and gonadrotropins are low.

Published Books

- Pediatric Clinical Diagnosis
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- Basic Pediatric Radiology
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UNIVERSITY BOOK CENTRE
8 Soliman El Halaby Street., Cairo
Tel. 5774881 - 3957807
Fax: 5897635 - Mobile 0123698600